# ALOX12B gene

arachidonate 12-lipoxygenase, 12R type

## Normal Function

The *ALOX12B* gene provides instructions for making an enzyme called 12R-LOX. This enzyme is part of a family of enzymes called arachidonate lipoxygenases. Most of these enzymes help add an oxygen molecule to a certain fatty acid called arachidonic acid. The addition of an oxygen molecule to arachidonic acid produces substances called fatty acid hydroperoxides, which can be transformed into a variety of signaling molecules. Specifically, the 12R-LOX enzyme helps add an oxygen molecule to arachidonic acid to make a substance called 12R-hydroperoxyeicosatetraenoic acid (12R-HPETE). 12R-HPETE is later converted to a signaling molecule that is involved in the growth and division (proliferation) and specialization (differentiation) of skin cells.

The 12R-LOX enzyme is thought to play a role in the formation and maintenance of the fat (lipid) membrane of the cells that make up the outermost layer of the skin (the epidermis). The epidermis helps prevent water loss, regulates body temperature, and protects against infection.

## Health Conditions Related to Genetic Changes

### nonbullous congenital ichthyosiform erythroderma

More than 30 mutations in the *ALOX12B* gene have been found to cause nonbullous congenital ichthyosiform erythroderma (NBCIE). Most of these mutations change single protein building blocks (amino acids) in the 12R-LOX enzyme. Many *ALOX12B* gene mutations lead to the production of a nonfunctional 12R-LOX enzyme, which impairs the formation of the lipid membrane of the cells within the epidermis. Problems with this protective barrier underlie the skin abnormalities and other features of NBCIE.

### other disorders

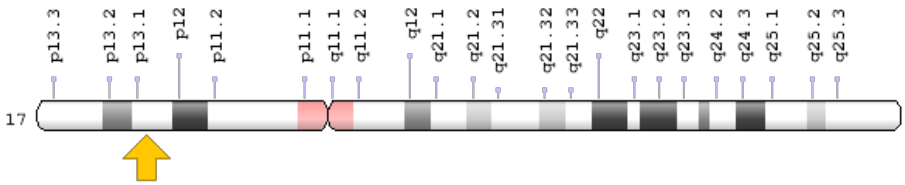
Another form of ichthyosis called self-healing collodion baby has been found to be caused by *ALOX12B* gene mutations. Individuals with this condition are born with a tight, clear sheath covering their skin called a collodion membrane. This membrane is usually shed during the first few weeks of life, and affected infants often show near normal skin within a few months.

Only a few people diagnosed with self-healing collodion baby have been found to have *ALOX12B* gene mutations; the majority of cases are caused by mutations in other genes.

## Chromosomal Location

Cytogenetic Location: 17p13.1, which is the short (p) arm of chromosome 17 at position 13.1

Molecular Location: base pairs 8,072,636 to 8,087,703 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- 12R-lipoxygenase
- 12R-LOX
- arachidonate 12-lipoxygenase, 12R-type
- epidermis-type lipoxygenase 12
- LX12B_HUMAN

## Additional Information & Resources

GeneReviews

- Autosomal Recessive Congenital Ichthyosis
  https://www.ncbi.nlm.nih.gov/books/NBK1420

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ALOX12B%5BTIAB%5D%29+OR+%2812R-LOX%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D

OMIM

- ARACHIDONATE 12-LIPOXYGENASE, R TYPE
  http://omim.org/entry/603741

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ALOX12B.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ALOX12B%5Bgene%5D
- HGNC Gene Family: Arachidonate lipoxygenases
  http://www.genenames.org/cgi-bin/genefamilies/set/407
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=430
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/242
- UniProt
  http://www.uniprot.org/uniprot/O75342

## Sources for This Summary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